



myosin storage myopathy

Myosin storage myopathy is a condition that causes muscle weakness (myopathy) that does not worsen or worsens very slowly over time. This condition is characterized by the formation of protein clumps, which contain a protein called myosin, within certain muscle fibers. The signs and symptoms of myosin storage myopathy usually become noticeable in childhood, although they can occur later. Because of muscle weakness, affected individuals may start walking later than usual and have a waddling gait, trouble climbing stairs, and difficulty lifting the arms above shoulder level. Muscle weakness also causes some affected individuals to have trouble breathing.

Frequency

Myosin storage myopathy is a rare condition. Its prevalence is unknown.

Genetic Changes

Mutations in the *MYH7* gene cause myosin storage myopathy. The *MYH7* gene provides instructions for making a protein known as the cardiac beta (β)-myosin heavy chain. This protein is found in heart (cardiac) muscle and in type I skeletal muscle fibers, one of two types of fibers that make up the muscles that the body uses for movement. Cardiac β -myosin heavy chain is the major component of the thick filament in muscle cell structures called sarcomeres. Sarcomeres, which are made up of thick and thin filaments, are the basic units of muscle contraction. The overlapping thick and thin filaments attach to each other and release, which allows the filaments to move relative to one another so that muscles can contract.

Mutations in the *MYH7* gene lead to the production of an altered cardiac β -myosin heavy chain protein, which is thought to be less able to form thick filaments. The altered proteins accumulate in type I skeletal muscle fibers, forming the protein clumps characteristic of the disorder. It is unclear how these changes lead to muscle weakness in people with myosin storage myopathy.

Inheritance Pattern

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

Other Names for This Condition

- autosomal dominant hyaline body myopathy

Diagnosis & Management

These resources address the diagnosis or management of myosin storage myopathy:

- Genetic Testing Registry: Myosin storage myopathy
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1842160/>

These resources from MedlinePlus offer information about the diagnosis and management of various health conditions:

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care
<https://medlineplus.gov/palliativecare.html>

Additional Information & Resources

MedlinePlus

- Encyclopedia: Muscle Function Loss
<https://medlineplus.gov/ency/article/003190.htm>
- Health Topic: Muscle Disorders
<https://medlineplus.gov/muscledisorders.html>

Genetic and Rare Diseases Information Center

- Myosin storage myopathy
<https://rarediseases.info.nih.gov/diseases/7148/myosin-storage-myopathy>

Additional NIH Resources

- National Institute of Neurological Disorders and Stroke: Congenital Myopathy Information Page
<https://www.ninds.nih.gov/Disorders/All-Disorders/Congenital-Myopathy-Information-Page>

Educational Resources

- MalaCards: myosin storage myopathy
http://www.malacards.org/card/myosin_storage_myopathy
- Muscular Dystrophy Association: Facts About Myopathies
https://www.mda.org/sites/default/files/publications/Facts_Myopathies_P-208.pdf
- New York Presbyterian Hospital: Myopathy
<http://www.nyp.org/neuro/services/neuromuscular-disorders/myopathy>

Patient Support and Advocacy Resources

- Muscular Dystrophy Association
<https://www.mda.org/>
- Muscular Dystrophy UK
<http://www.musculardystrophyuk.org/>

Genetic Testing Registry

- Myosin storage myopathy
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1842160/>

ClinicalTrials.gov

- ClinicalTrials.gov
<https://clinicaltrials.gov/ct2/results?cond=%22myosin+storage+myopathy%22>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28myosin+storage+myopathy%5BTIAB%5D%29+OR+%28autosomal+dominant+hyaline+body+myopathy%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- MYOPATHY, MYOSIN STORAGE, AUTOSOMAL DOMINANT
<http://omim.org/entry/608358>

Sources for This Summary

- Armel TZ, Leinwand LA. Mutations in the beta-myosin rod cause myosin storage myopathy via multiple mechanisms. *Proc Natl Acad Sci U S A*. 2009 Apr 14;106(15):6291-6. doi: 10.1073/pnas.0900107106. Epub 2009 Mar 31.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/19336582>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2669361/>
- Pegoraro E, Gavassini BF, Borsato C, Melacini P, Vianello A, Stramare R, Cenacchi G, Angelini C. MYH7 gene mutation in myosin storage myopathy and scapulo-peroneal myopathy. *Neuromuscul Disord*. 2007 Apr;17(4):321-9. Epub 2007 Mar 2.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/17336526>
- Tajsharghi H, Oldfors A. Myosinopathies: pathology and mechanisms. *Acta Neuropathol*. 2013 Jan; 125(1):3-18. doi: 10.1007/s00401-012-1024-2. Epub 2012 Aug 5. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/22918376>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3535372/>
- Tajsharghi H, Thornell LE, Lindberg C, Lindvall B, Henriksson KG, Oldfors A. Myosin storage myopathy associated with a heterozygous missense mutation in MYH7. *Ann Neurol*. 2003 Oct; 54(4):494-500.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/14520662>

Reprinted from Genetics Home Reference:

<https://ghr.nlm.nih.gov/condition/myosin-storage-myopathy>

Reviewed: February 2013

Published: February 14, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services